

Masters

Lesson 1—What Is a Rare Disease?

Activity 1: What Is a Rare Disease?

- Master 1.1, *Letter from a Producer* transparency
Master 1.2, *Thinking about Disease*..... student copies

Lesson 2—What Causes Rare Diseases?

Activity 1: Causes of Disease

- Master 2.1, *Medical Officer Report Form* transparency and student copies
for each pair
Master 2.2, *Visits to the Infirmary, Week 1** student copies for each pair
Master 2.3, *Test Results, Week 1** student copies for each pair

Activity 2: Is a Rare Disease Present?

- Master 2.4, *Follow-up on Week 1 Infirmary Visits** student copies for each pair
Master 2.5, *Visits to the Infirmary, Week 2** student copies for each pair
Master 2.6, *Medical Reference Manual: Necrotizing Fasciitis** student copies for each pair
Master 2.7, *Questions about a Rare Disease* student copies for each pair

Lesson 3—The Difficulty of Diagnosis

Activity 1: A Parent's Dilemma

- Master 3.1, *To Play or Not to Play?* transparency
Master 3.2, *Medical Specialty Report Form* student copies
Master 3.3, *Heart and Circulatory System** student copies for each group of 4
Master 3.4, *Vision System** student copies for each group of 4
Master 3.5, *Respiratory System** student copies for each group of 4
Master 3.6, *Skeletal System** student copies for each group of 4

Activity 2: Connective Tissue

- Master 3.7, *Measuring Elasticity* transparency

Activity 3: A Common Thread

- Master 3.8, *Diagnosing a Connective Tissue Disorder*. student copies for each pair
Master 3.9, *Medical Reference Manual: Disorders
of the Connective Tissue** student copies for each pair
Master 3.10, *Patrick's Family** student copies for each pair
Master 3.11, *Living with Marfan Syndrome** transparency

Lesson 4—The Importance of Medical Research

Activity 1: An Unwelcome Diagnosis

- Master 4.1, *Doctor Visits 1 and 2* transparency
Master 4.2, *Internet Search Results*..... student copies

*Print version only

Master 4.3, <i>Evaluating Internet Search Results</i>	transparency and student copies
Master 4.4, <i>Summarizing Information about Leukemia</i>	student copies for each pair
Master 4.5, <i>Karyotype</i>	student copies

Activity 2: Clinical Trials

Master 4.6, <i>Another Doctor Visit</i>	transparency
Master 4.7, <i>Treating Leukemia</i>	student copies for each pair
Master 4.8, <i>Designing a Clinical Trial</i>	student copies for each pair
Master 4.9, <i>Survival Rates for Children with Leukemia</i>	student copies for each pair
Master 4.10, <i>Information about Leukemia*</i>	student copies for each pair
Master 4.11, <i>Family Karyotypes*</i>	student copies
Master 4.12, <i>Hanna's Karyotypes*</i>	student copies
Master 4.13, <i>Changing the Dose of Drug A*</i>	student copies for each pair
Master 4.14, <i>Changing the Dose of Drug B*</i>	student copies for each pair
Master 4.15, <i>Changing the Dose of Drug C*</i>	student copies for each pair
Master 4.16, <i>Central Nervous System Treatment*</i>	student copies for each pair
Master 4.17, <i>Interview with Hailey*</i>	2 copies for the class

Lesson 5—Communicating about Rare Diseases

Activity 1: Creating an Informational Poster

Master 5.1, <i>Guidelines for the Poster</i>	transparency and student copies for each pair
Master 5.2, <i>Research Study on Marfan Syndrome</i>	student copies for each pair
Master 5.3, <i>Clinical Trial on Childhood Leukemia</i>	student copies for each pair
Master 5.4, <i>Evaluation Rubric for Poster</i>	transparency and student copies for each pair
Master 5.5, <i>Poster Score Sheet</i>	student copies for each pair

Activity 2: Reflecting on Rare Diseases

Master 5.6, <i>What Do You Think Now?</i>	student copies
Master 5.7, <i>Another Letter from the Producer*</i>	transparency

*Print version only

Letter from a Producer



Dear Principal:

I am a producer for People Reality Productions, a company that develops reality television shows for broadcast over several cable TV channels. We are thinking about creating a series that follows the life of a student who has a rare disease.

We are still working out the details of the series. At this point, we haven't identified the student who will participate or even which rare disease will be featured in the series. As a first step, we are interested in learning what concerns and questions your teachers and students have about a student with a rare disease joining their class.

Please share this letter with your teachers. After they have had a chance to discuss it with their students, I will call you to learn about their thoughts and concerns.

Thank you for your help with this project.

Sincerely,

Vincent Shifflett
Senior Producer
People Reality Productions

Thinking about Disease

Name: _____

Questions

1. What is a disease?
2. How do doctors tell if someone has a disease?
3. What do you think causes disease?
4. Make a list of list 10 different diseases.

Table 1. List of Diseases

5. What does it mean to call a disease “rare”?
6. In simple terms, curing a disease means that the patient has been restored to good health and there is little chance of the disease coming back.

It is not possible to cure all diseases. Controlling a disease means that the disease symptoms are lessened and the quality of the life for the patient is improved, but the disease has not been cured.

Which of the diseases you listed in Step 4 do you think are curable, controllable, or incurable?

Medical Officer Report Form

Name(s): _____

Questions

1. Summarize the reasons that soldiers came to the infirmary during Week 1.
2. Fill out the form for ordering lab tests.

Test Order Form

Soldier	Test ordered	Why was the test ordered?
1		
2		
3		
4		
5		
6		
7		
8		
9		
10		
11		
12		
13		
14		
15		
16		
17		
18		
19		
20		
21		

Visits to the Infirmary, Week 1

Soldier	Barracks	What's wrong?	Test ordered	Test results	Diagnosis	Treatment
1	A	Sore, red, itchy eyes				
2	D	Sore toe				
3	A	Sore, red, itchy eyes				
4	A	Sore, red, itchy eyes				
5	G	Sore throat				
6	A	Sore, red, itchy eyes				
7	E	Skin rash				
8	A	Sore, red, itchy eyes				
9	F	Cut on left leg				
10	G	Sore throat				
11	I	Shortness of breath				
12	A	Sore, red, itchy eyes				
13	H	Sore toe				
14	G	Sore throat				
15	E	Skin rash				
16	G	Sore throat				
17	G	Sore throat				
18	B	Ankle pain				
19	G	Sore throat				
20	D	Sore toe				
21	C	Sore toe				

Test Results, Week 1

Soldier	Barracks	What's wrong?	Test ordered	Test results	Diagnosis	Treatment
1	A	Sore, red, itchy eyes	Test 1	+ for bacteria C	Pinkeye	Antibiotic eyedrops
2	D	Sore toe	Test 1	+ for bacteria A	Blister from boots	Dressing
3	A	Sore, red, itchy eyes	Test 1	+ for bacteria C	Pinkeye	Antibiotic eyedrops
4	A	Sore, red, itchy eyes	Test 1	+ for bacteria C	Pinkeye	Antibiotic eyedrops
5	G	Sore throat	Test 1	+ for bacteria A	Strep throat	Antibiotic
6	A	Sore, red, itchy eyes	Test 1	+ for bacteria C	Pinkeye	Antibiotic eyedrops
7	E	Skin rash	Test 2	+ for poison ivy	Poison ivy	Steroid skin cream
8	A	Sore, red, itchy eyes	Test 1	+ for bacteria C	Pinkeye	Antibiotic eyedrops
9	F	Cut on left leg	No test			Stitches
10	G	Sore throat	Test 1	+ for bacteria A	Strep throat	Antibiotic
11	I	Shortness of breath	No test		Asthma	Inhaler
12	A	Sore, red, itchy eyes	Test 1	+ for bacteria C	Pinkeye	Antibiotic eyedrops
13	H	Sore toe	Test 1	+ for bacteria A	Blister from boots	Dressing
14	G	Sore throat	Test 1	+ for bacteria A	Strep throat	Antibiotic
15	E	Skin rash	Test 2	+ for poison ivy	Poison ivy	Steroid skin cream
16	G	Sore throat	Test 1	+ for bacteria A	Strep throat	Antibiotic
17	G	Sore throat	Test 1	+ for bacteria A	Strep throat	Antibiotic
18	B	Ankle pain	No test	X-ray	No break	Bandage
19	G	Sore throat	Test 1	+ for bacteria A	Strep throat	Antibiotic
20	D	Sore toe	Test 1	– for all bacteria	Blister from boots	Dressing
21	C	Sore toe	Test 1	+ for bacteria A	Blister from boots	Dressing

Follow-up on Week 1 Infirmary Visits

Summary of Patients' Responses to Treatment from Week 1

- Six soldiers from Barracks A with pinkeye tested positive for infection by bacterial species C. They were treated with eyedrops containing antibiotics.
In all cases, the infection has cleared up.
- Six soldiers from Barracks G with sore throats tested positive for infection by bacterial species A. They were treated with antibiotics. Although two soldiers returned to the infirmary, all are responding to the antibiotics and the infections have largely disappeared.
- Four soldiers developed blisters from wearing new boots. Three of the four tested positive for infection by bacterial species A, and the fourth soldier tested negative for species A, B, and C. One soldier received a cut on the left leg, which was closed with stitches. He has no evidence of infection.
- One soldier twisted his ankle. X-rays showed no broken bones. The ankle was bandaged, and the soldier has been assigned to light duty.
- Two soldiers from Barracks E tested positive for exposure to poison ivy. They were treated with steroid cream, and the skin rashes are disappearing.
- One soldier, who was short of breath, was diagnosed as having asthma and was given an inhaler, which eased her symptoms.

Visits to the Infirmary, Week 2

Return Visits to the Infirmary from Week 1 Soldier Visits

Soldier	Barracks	What's wrong?	Tests ordered	Test results	Diagnosis	Treatment
2	D	Swollen, blistered lower leg				
16	G	Sore throat				
19	G	Sore throat				

New Visits to the Infirmary, Week 2

Soldier	Barracks	What's wrong?	Tests ordered	Test results	Diagnosis	Treatment
22	G	Sore throat				
23	A	Sore throat				
24	E	Cut on head				
25	G	Sore throat				
26	G	Sore throat				
27	B	Skin rash				
28	G	Sore throat				
29	D	Ankle pain				
30	A	Sore throat				
31	B	Sore toe				
32	F	Skin rash				

Medical Reference Manual: Necrotizing Fasciitis (Flesh-Eating Bacteria)

What is it?

Flesh-eating disease is a bacterial infection that destroys skin and fat tissue. The disease is very rare. The odds of getting it are about 1 in 100,000. However, it is very serious. About 2 out of 10 people who get this infection die from it.

What causes it?

The disease can be caused by different species of bacteria, including the one that causes strep throat. The bacteria enter the body through open wounds, where they interact with the immune system to produce the disease. Flesh-eating disease is rare because the immune system of most people will stop the infection before it becomes serious.

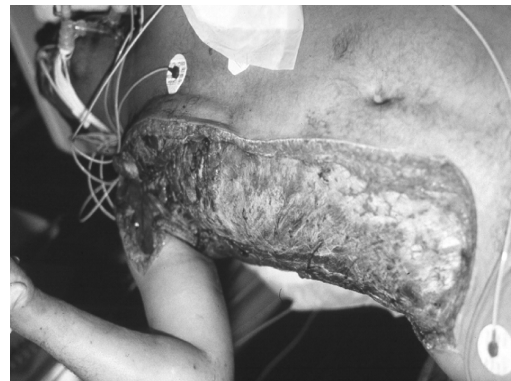
What are the symptoms?

The skin reddens, becomes swollen, and is painful to the touch. Other symptoms include nausea, vomiting, and diarrhea. The symptoms start suddenly, may get better for a day or two, then quickly worsen. If not treated, the disease may result in organ failure and death.

Figure 1. Early infection. (Donald E. Low, University Health Network/Mount Sinai Hospital)



Figure 2. Late infection. (Donald E. Low, University Health Network/Mount Sinai Hospital)



How is it treated?

Patients with flesh-eating disease need immediate hospital care. Treatment involves antibiotics and surgery to remove diseased tissue and stop the spread of the disease.

Questions about a Rare Disease

Name(s): _____

1. What evidence suggests that bacterial species A causes both sore throat and foot infection?
2. What evidence suggests that the soldier with the foot infection has flesh-eating disease?
3. Why are there many cases of sore throat but only one case of flesh-eating disease?
4. What evidence is there that flesh-eating disease is a rare disease?
5. What should be the next step in treating the soldier with the foot infection? Explain your reasoning.

To Play or Not to Play?

Patrick is a 13-year-old middle school student who loves to play basketball. He came home excited from school and explained to his parents that the school basketball team will be holding tryouts next month and he wants to participate.

Patrick's parents are both happy and concerned for him. They are happy because they know Patrick loves sports, and they feel that the exercise will be good for him. They also know that Patrick has been occasionally teased because he is tall and thin. Maybe by joining the basketball team he will make new friends and feel more accepted by his classmates.

Patrick's parents are concerned because he has some health problems. When Patrick was a toddler, the family doctor diagnosed him with a heart murmur. The doctor explained that a heart murmur refers to a sound that the blood makes as it flows through the heart. She further explained that heart murmurs are usually harmless and that Patrick could lead a normal life.

When he was nine, Patrick developed a problem with his eyesight, and it was discovered that one of his eye lenses was detached and had to be repaired. When Patrick was 10, he was diagnosed with asthma. The doctor explained that asthma causes the tubes carrying air in and out of the lungs to become sore and swollen. This can cause coughing and wheezing and make it difficult to breathe. The doctor created a treatment plan for Patrick that helped him recognize his symptoms and use an inhaler to make breathing easier. She also explained that, with proper management of his asthma, Patrick could play sports and that the exercise might even improve his condition.

Finally, just last year, Patrick was diagnosed with scoliosis, or curvature of the spine. The doctor explained that Patrick's scoliosis was moderate and, as with most children, the cause was unknown. He further explained that in 90 percent of cases, no future treatment is needed.

Medical Specialty Report Form

Name(s): _____

Patient's name _____

Medical specialty _____

Patient's medical history

--

Results from physical exam

--

Possible causes

--

Heart and Circulatory System

Cardiologist Report

Medical history

Patient was previously diagnosed with a heart murmur. An echocardiogram reveals mitral valve prolapse and an enlarged aorta.

Physical exam

The presence of a heart murmur was confirmed. An echocardiogram revealed the presence of mitral valve prolapse.

Medical Reference Manual: *Heart and Circulatory System*

Heart murmur

When doctors use a stethoscope to listen to the heartbeat, they hear a lub-DUB sound made by heart valves opening and closing as blood flows through the heart. The term “heart murmur” refers to an unusual whooshing sound doctors hear when listening to the heartbeat.

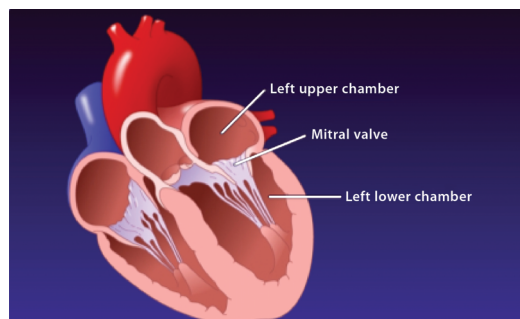
Doctors diagnosis heart murmurs in many children at some point in their lives. Most heart murmurs are harmless and need no treatment. Other heart murmurs are called abnormal and may be associated with defects in the heart that were present at birth.

Mitral valve prolapse

In the condition called mitral valve prolapse, one of the heart’s valves doesn’t work properly. The valve flaps are “floppy” and don’t close properly. This sometimes causes blood to flow backward from its normal direction. This backflow of blood may be associated with shortness of breath or chest pain.

The cause of mitral valve prolapse is not known. Most people with the condition are born with it. It tends to run in families and is associated with connective tissue disorders such as Marfan syndrome.

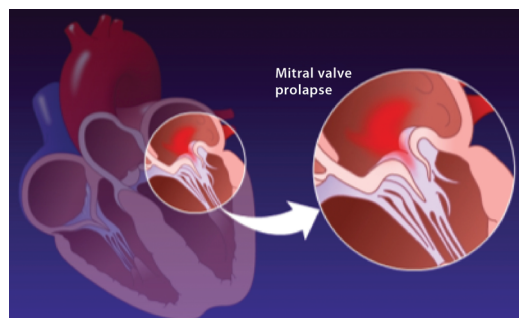
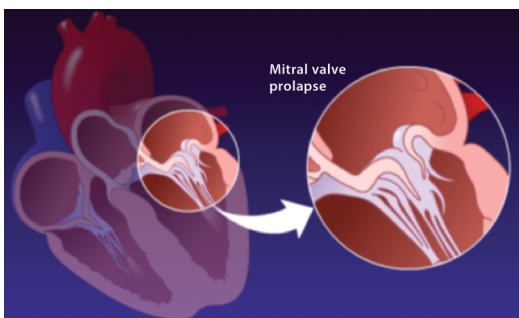
Figure 1. *Mitral valve prolapse.*



The mitral valve is a valve that lies between the left upper and lower chambers of the heart.

In mitral valve prolapse, the valve flaps are too large and don’t form a tight seal when they close.

This lack of a tight seal can cause a small amount of blood to flow backward, resulting in a heart murmur.



Vision System

Ophthalmologist Report

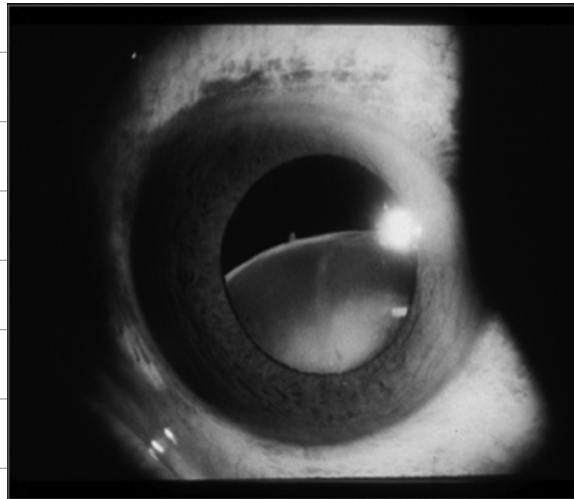
Medical history

The patient is myopic (nearsighted). When he was nine years old, he was being fitted for eyeglasses when an exam revealed that his left lens was dislocated.

Physical exam

An eye exam confirmed myopia and a repaired detached left lens.

Figure 1. Slit lamp exam: Photo from a slit lamp exam of a patient's eye showing a detached left lens. (Kevin J. Blinder, MD, The Retina Institute, Washington University School of Medicine)



Medical Reference Manual: *Vision System*

Myopia (nearsightedness)

Nearsightedness is caused by a change in the shape of the eyeball so that it is egg shaped instead of round. This egg-shaped eyeball focuses light a little in front of the retina instead of directly on it, resulting in blurry vision. Myopia is a common condition affecting 30 to 40 percent of the American population.

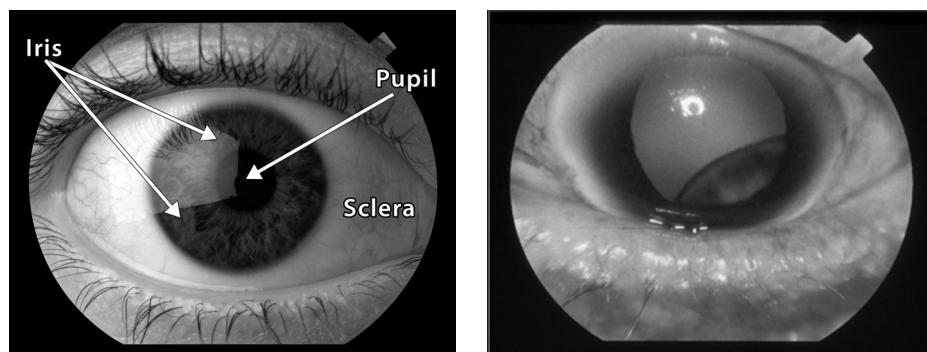
Detached lens

A detached, or dislocated, lens means that the lens has shifted from its normal position (centered behind the pupil). If the dislocation is moderate, the problem may be corrected with glasses. A severe lens dislocation may require surgery to correct.

Detached lenses are rare in the general population. They are often caused by a blow to the eye. The condition is much more common among people with certain diseases involving connective tissue such as Ehlers-Danlos syndrome and Marfan syndrome.

Figure 2. Photos from slit lamp exams: Left, a normal lens; right, a detached lens.

((left) Corbis, (right) Kevin J. Blinder, MD, The Retina Institute, Washington University School of Medicine)



Respiratory System

Pulmonologist Report

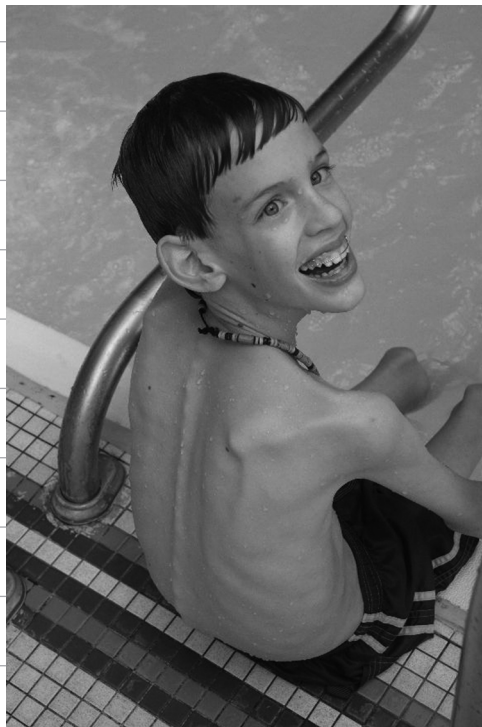
Medical history

Patient has been diagnosed with asthma. When seven years old, he experienced a collapsed lung.

Physical exam

Exam confirmed the diagnosis of asthma. Lung volume was normal. Chest X-ray was normal.

Figure 1. Patient photo: Patrick at age 10.



Medical Reference Manual: *Respiratory System*

Table 1. Information about Asthma

What are the symptoms of asthma?	<ul style="list-style-type: none">• Coughing• Wheezing• Difficulty breathing
What causes asthma attacks?	<p>Asthma attacks are a response to environmental triggers that leads to</p> <ul style="list-style-type: none">• Inflammation of the airways. This swelling and irritation inside the airways leads to difficulty breathing.• Bronchospasms. The muscles surrounding the airways go into spasm, leading to their narrowing. <p>In rare cases, asthma is associated with disorders of the connective tissue.</p>
What are the triggers for asthma attacks?	<ul style="list-style-type: none">• Dust• Mold• Pollen• Pets• Cigarette smoke• Pollution• Illness (infection by bacteria and viruses)
How is asthma treated?	<p>Inhaled drugs, such as albuterol, help widen the airways during asthma attacks and make it easier to breathe.</p> <p>For long-term management of asthma, inhaled steroids are safe and can be used every day.</p>

Skeletal System

Orthopedist Report

Medical history

Patient was previously diagnosed with mild scoliosis (curvature of the spine).

Physical exam

A physical exam and X-rays confirmed the presence of mild scoliosis. The curvature was measured to 15 degrees. It was noted that the patient has unusually long, slender arms, fingers, and feet.

Figure 1. Patient X-ray: X-ray taken when patient was five years old.

(© Ldambies | Dreamstime.com)



Medical Reference Manual: *Skeletal System*

Scoliosis (curvature of the spine)

Scoliosis refers to an abnormal curvature of the spine. About 10 percent of adolescents show some degree of scoliosis, but less than 1 percent need treatment for the condition. The severity of scoliosis is described by the extent of the curvature.

Table 1. Severity of Scoliosis

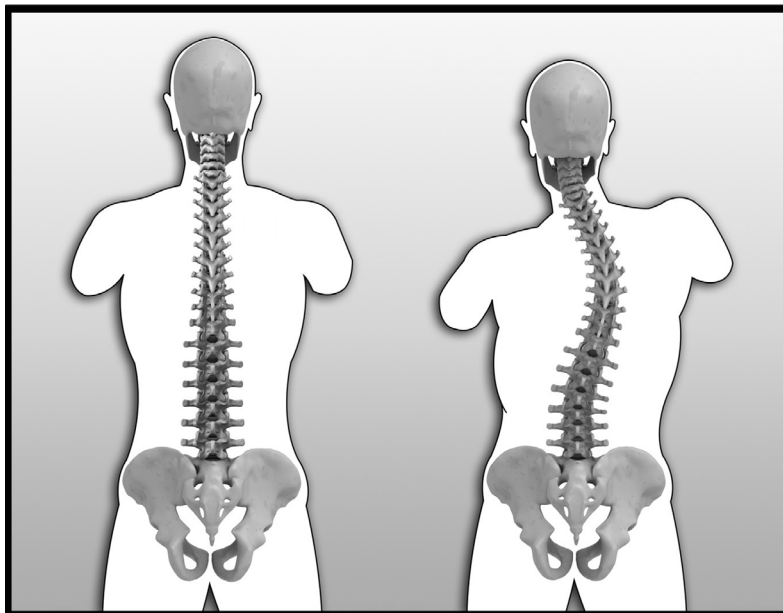
Amount of Curvature	Severity
Curvature less than 20 degrees	Mild
Curvature between 20 and 70 degrees	Moderate
Curvature greater than 70 degrees	Severe

Patients with mild scoliosis usually don't require treatment beyond examination to see whether the condition worsens. Patients with moderate and severe scoliosis are treated with back braces or surgery.

In most cases, the cause of scoliosis is not known; however, it does seem to run in families. In some cases, the condition is caused by an injury. In other cases, the condition is a result of a muscle, nerve, or connective tissue disease.

Figure 2. The spine and scoliosis: Left, normal spine; right, spine showing scoliosis.

(© Sebastian Kaulitzki | Dreamstime.com)

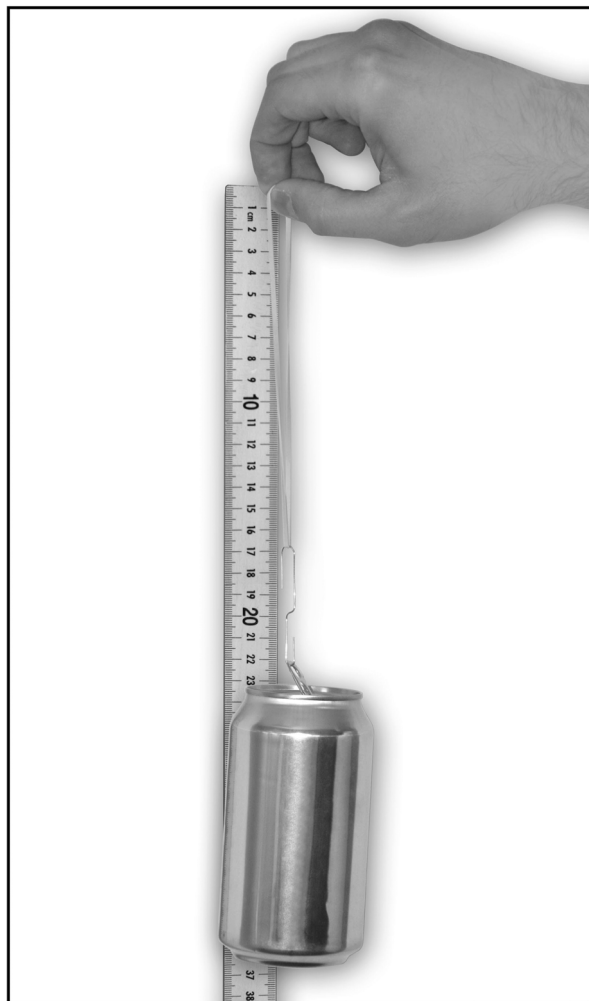


Measuring Elasticity

Steps for measuring the elasticity of rubber bands

1. Unfold the paper clip into an S shape.
2. Slip one end of the paper clip through the pulled tab on top of the soda can.
3. Place the other end of the paper clip through the rubber band.
4. Hold a meter stick upright on a hard surface so that the end reading “100 centimeters” is resting on the surface and the end reading “0 centimeters” is up in the air.
5. Hold the top of rubber band (the end away from the paper clip) up to the end of the meter stick that reads “0.”
6. Observe and record in your notebook how far down the meter stick the rubber band has stretched.
7. Repeat Steps 2 through 6 for the second rubber band.

Figure 1. Measuring elasticity.



Diagnosing a Connective Tissue Disorder

Name(s): _____

1. You will try to match Patrick's medical symptoms to four different disorders of connective tissue.
2. For each of Patrick's symptoms listed in the table under "medical history," decide whether that symptom is consistent with each of the four connective tissue disorders written across the top row. Use the information supplied in the Medical Reference Manual.
3. Place a check mark in the appropriate box when the symptom is consistent with the connective tissue disorder.

Table 1. Checklist of Patrick's Symptoms

Patrick's medical history	Ehlers-Danlos syndrome	Marfan syndrome	Osteogenesis imperfecta	Scleroderma
Myopia				
Detached eye lens				
Asthma				
Collapsed lung				
Heart murmur				
Leaky heart valve				
Long arms and legs				
Curvature of spine				

Medical Reference Manual: Disorders of the Connective Tissue

Connective tissues are made of proteins and fats. They support your body's organs and give your tissues their shape. Cartilage is an important connective tissue. It is stiff but more flexible than bone. Cartilage helps your bones move and glide over each other. It also gives shape to body parts such as your nose and ears.

Connective tissue may be damaged by injury or through an infection. It can also be damaged by a large number of genetic disorders that occur rarely in the population. A few of them are described below.

Ehlers-Danlos syndrome

Ehlers-Danlos syndrome refers to a collection of related disorders that weaken connective tissues. Symptoms can be mild to life threatening. They include the following:

- heart valves that leak
- weakened blood vessels
- loose joints
- abnormal wound healing
- soft, stretchy skin that bruises easily
- muscle weakness
- joint dislocations

Ehlers-Danlos syndrome is an inherited disorder. Treatment involves managing symptoms and learning how to protect the joints and prevent injuries.

Scleroderma

Scleroderma is a group of related disorders involving abnormal growth of connective tissue. One type of scleroderma affects only the skin. Another type can also affect other body systems. The cause of scleroderma is not known. It is more common in females than males. Other symptoms may include the following:

- calcium deposits in connective tissues
- narrowing of blood vessels in the hands and feet
- swelling of the esophagus (tube between the throat and stomach)
- thick, tight skin on fingers
- red spots on hands and face

Treatment involves managing the symptoms.

Marfan syndrome

Marfan syndrome is a disorder of connective tissue that is due to mutations in a gene that codes for a connective tissue protein called fibrillin. Symptoms can be mild to severe. Often, people with Marfan syndrome are tall and thin and have loose joints. Their fingers and feet may be unusually long. Other symptoms may include the following:

- heart valves that leak
- heart murmur
- weakened blood vessels
- curvature of the spine
- flat feet
- sudden lung collapse, sometimes asthma
- nearsightedness and problems with the eye lens
- stretch marks on the skin
- teeth that are crowded together

Marfan syndrome is an inherited disorder. Treatment involves managing symptoms and adopting physical activity guidelines that are specific to each person.

Osteogenesis imperfecta

Osteogenesis imperfecta is an inherited disorder that causes bone weakness. The disorder is caused by mutations to a gene involved with making the protein collagen. Sometimes, bones break for no apparent reason. Symptoms can be mild to severe. Other symptoms may include the following:

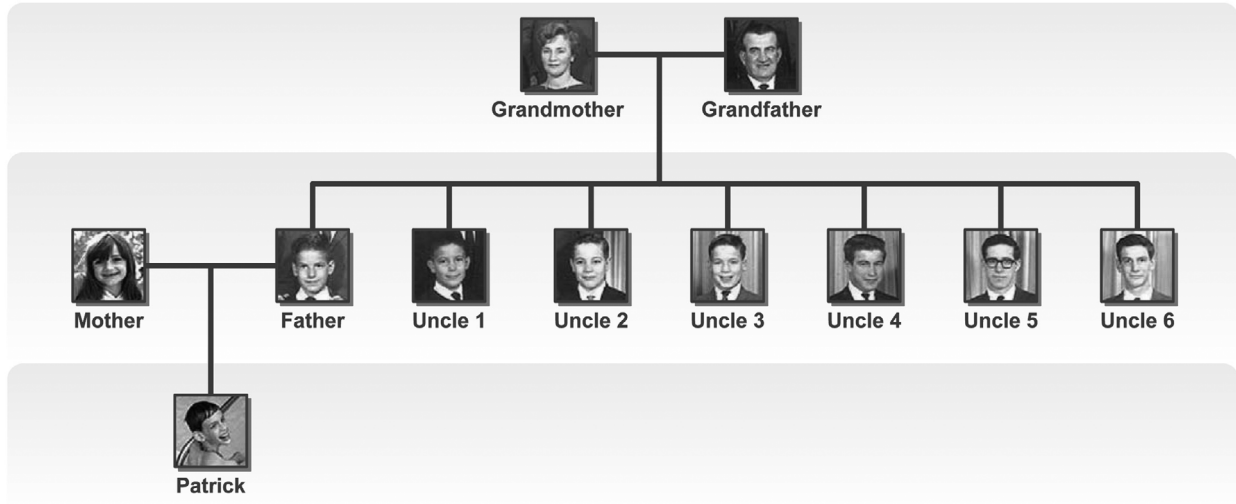
- muscle weakness
- curvature of the spine
- loose joints
- hearing loss
- skin that bruises easily
- brittle teeth

Treatment involves exercise, physical therapy, braces, and surgery.

Patrick's Family

Figure 1. Patrick's family on his father's side.

Patrick's Family Tree



Grandmother: In good health

Grandfather: Died after several heart operations

Mother: In good health

Father: Has had two heart operations and has mild scoliosis

Uncle 1: Died from brain cancer

Uncle 2: In good health

Uncle 3: In good health

Uncle 4: Has had a heart operation and a collapsed lung

Uncle 5: Died after several heart operations and had a detached eye lens

Uncle 6: Has type 2 diabetes

Living with Marfan Syndrome

Questions to a doctor from young people who have Marfan syndrome

1. “Is there any possible way for the Marfan gene to be detected before a child is born and maybe find a way to prevent it from mutating itself?”
2. “When they measured my heart with the echocardiogram, they told my mom they don’t think I should do marching band. I was wondering, if I don’t exert myself too much, if I take it at my own pace, do you think I could do it?”

Comments from those young people about the most frustrating part of having Marfan syndrome

1. “The most frustrating thing for me is ... I can’t drive. I tried to get my permit, and I couldn’t pass the vision test because I did have my retina detached.”
2. “The thing that frustrates me the most is all the aches and pains in my joints and sternum.”
3. “When it comes to how you’re socially accepted, high school is really lame. In a couple of years, it’s not going to matter what sport you played or anything. It is going to matter what you know and what you do with the knowledge that you know.”

Doctor Visits 1 and 2

Background

Jason and Kim are the parents of a five-year old girl, Hanna. They are concerned because she has had flu-like symptoms for three weeks and has not responded to treatment. The family doctor was concerned that something more serious than flu might be responsible for Hanna's symptoms. He referred Hanna to another doctor for further examination.

First visit

The doctor examined the child and immediately noticed that she had signs of an infection. She felt the child's abdomen and observed that the liver appeared to be swollen. The doctor asked the parents about their daughter's health and the health of the rest of the family. Finally, she ordered some blood tests.

Second visit

During the follow-up visit, the doctor explained that the results of the blood tests showed there was a problem. The doctor explained that Hanna had developed a cancer of the blood called leukemia. Jason and Kim were understandably upset to hear this news. The doctor further explained the nature of the disease and how it would be treated. She told them that childhood leukemia such as Hanna's is treatable. About 80 percent of children with leukemia are cured after treatment.

Internet Search Results

1. **Federal Center for Cancer Research**

Information about leukemia, its causes, symptoms, diagnosis, and treatment ...

2. **My Leukemia Blog**

Living with Cancer: Reflections and remembrances of a cancer survivor ...

3. **The Cancer Research Center at Lincoln State University**

Breast cancer, Prostate cancer, Leukemia, Lymphoma, ...

4. **Information about Leukemia from the American Blood Cancer Society**

Cells of the blood, Stem cells and leukemia, White blood cells, bacteria ...

5. **Leukemia—Medhealthopedia: The Do-It-Yourself Encyclopedia**

Leukemia is a form of cancer that is ...

6. **Cancer drugs for less! Leukemia**

Order drugs from overseas to treat leukemia and save!

7. **Fed approves new drug to treat leukemia**

Medical Business Weekly (Washington, DC)—The Food and Drug Administration today approved Hamilton Pharmaceutical's drug Arresta for the treatment of leukemia ...

8. **Leukemia: Definition from medicaljargon.com**

Leukemia—A cancer of the white blood cells. White blood cells ...

Evaluating Internet Search Results

Name(s): _____

Questions

1. Hanna's parents searched the Internet because they wanted to find information about childhood leukemia. They wanted to answer such questions as
 - What causes leukemia?
 - What are its symptoms?
 - How is it diagnosed?
 - How is it treated?
 - How likely is it that the treatment will help?
2. When thinking about the results of an Internet search, for each hit, ask yourself:
 - Is this site likely to contain the information that I want?
 - Is this site likely to contain information that is accurate?
3. The Internet search returned the eight hits on your handout. Rank each hit in the table below by placing its number in one of the boxes. The most helpful hit is on the left side, and the least helpful hit is on the right side.

Table 1. Ranking the Hits

Most helpful				Least helpful			

Summarizing Information about Leukemia

Name(s): _____

Federal Center for Cancer Research

Disease definition

Disease symptoms

Disease diagnosis

American Blood Cancer Society

- List types and functions of blood cells.
- Where are blood cells made?
- What is a stem cell?
- What is the relationship between stem cells and leukemia?
- Why are people with leukemia more likely to get bacterial infections?

Karyotype

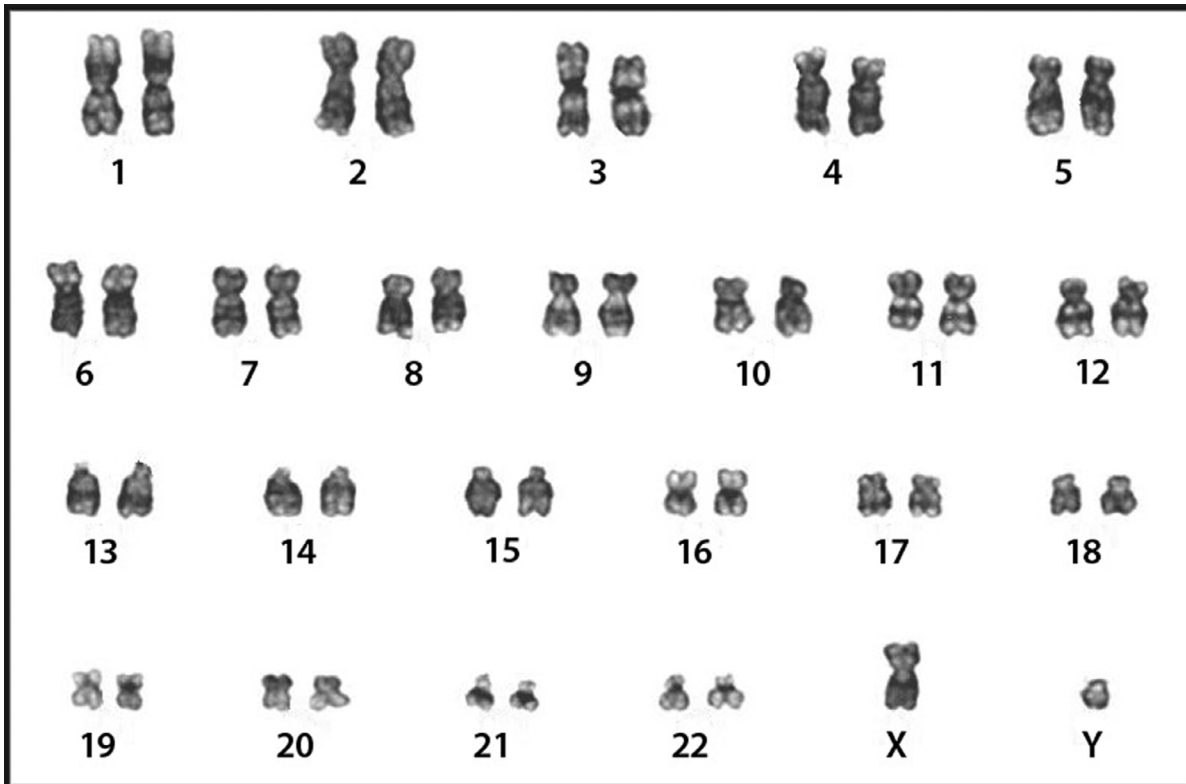
A karyotype is a photograph of a person's complete set of chromosomes.

A healthy human karyotype shows 22 pairs of numbered chromosomes (arranged by size), where one member of each pair comes from the mother and the other comes from the father.

There is an additional pair called the sex chromosomes. An individual with one X chromosome and one Y chromosome is male. Someone with two X chromosomes and no Y chromosome is female.

What is the sex of the person whose karyotype is above?

Figure 1. Sample karyotype. (Genetics Department, Affiliated Laboratories, Inc., Bangor, Maine.)



Another Doctor Visit

Jason and Kim returned to the doctor's office with Hanna to discuss her treatment. The doctor suggested having Hanna participate in something called a clinical trial. She explained that a clinical trial is a process in which groups of patients receive treatments that differ in one feature of their treatment.

Jason and Kim were alarmed at this suggestion. They asked,

- “Does this mean that you don't know what to do for her?”
- “Do you mean that Hanna may receive a sugar pill instead of a real drug?”

The doctor replied that for decades, most children with leukemia have participated in clinical trials. The results from these clinical trials have helped greatly improve survival rates. The doctor further explained that, during a clinical trial, patients are never given a sugar pill because effective treatment options are available. Instead, a control group of patients receives the standard therapy while another group of patients receives some experimental treatment.

Treating Leukemia

Treatments for childhood leukemia in 1970

Standard treatment: Chemotherapy

The standard treatment for childhood leukemia uses three drugs taken together. This combination of drugs and their doses have been guided by the results of many clinical trials:

- **Drug A is prednisone.** Cells have the ability to kill themselves if they become damaged. This helps keep the body free of unhealthy cells. Prednisone works by helping the body kill damaged white blood cells.
Side effects: Increased appetite, indigestion, and nervousness
- **Drug B is vincristine.** When cells divide, each new cell must receive an identical set of chromosomes. Vincristine interferes with this process and stops white blood cells from dividing.
Side effects: Hair loss, constipation, and nerve damage
- **Drug C is methotrexate.** When cells divide, they need to make more DNA, RNA, and protein molecules. Cancer cells are dividing rapidly compared with normal cells. Methotrexate interferes with the ability of all cells to make DNA, RNA, and proteins.
Side effects: Stomach pain, shortness of breath, and blood in urine

Central nervous system treatment

Doctors observed that often after chemotherapy treatment was stopped, leukemia reappeared in the central nervous system (brain and spinal cord). To kill cancer cells “hiding” in this part of the body, the patient’s head is exposed to X-rays and the drug methotrexate is injected directly into the spinal fluid.

Side effects: Learning problems and increased risk for heart disease

Designing a Clinical Trial

Name(s): _____

Clinical trial design

Drug A

- ☐ Decrease
- ☐ Standard dose
- ☐ Increase

Drug B

- ☐ Decrease
- ☐ Standard dose
- ☐ Increase

Drug C

- ☐ Decrease
- ☐ Standard dose
- ☐ Increase

Central nervous system treatment

- ☐ Yes
- ☐ No

Clinical trial results

Conclusion from the clinical trial

Survival Rates for Children with Leukemia

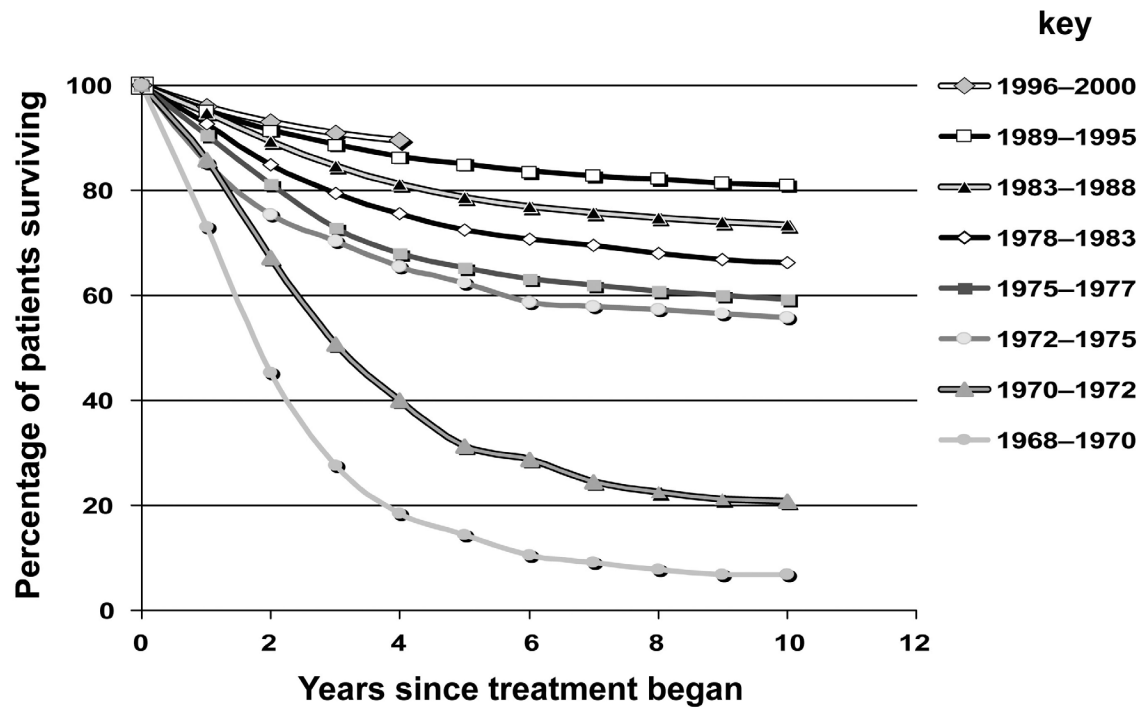


Image courtesy of Dr. Timothy Garrington

Information about Leukemia

Federal Center for Cancer Research

Leukemia: A cancer of the white blood cells. White blood cells associated with leukemia are abnormal, and they are produced in very high numbers. Although it is a rare disease, leukemia can affect both children and adults. One type of the disease can appear in a short period of time (days to weeks), while another type of the disease develops more slowly.

Disease symptoms catalog Leukemia

- Too few red blood cells and platelets
- Repeated infections by viruses and bacteria
- Pain in the joints
- Pain in the abdomen
- Wheezing and coughing

Diagnosing leukemia

Diagnosis of leukemia begins with a physical examination of the patient.

The doctor will

- look for signs of infection, such as runny nose, fever, and cough;
- feel the abdomen to see if the liver or spleen is enlarged;
- take a medical history that involves questions about
 - patient symptoms,
 - family health, and
 - medications and allergies; and
- order blood tests to measure the numbers of white blood cells, red blood cells, and platelets.

If the physical exam and blood test results suggest the possibility of leukemia, then the doctor may order the following tests:

- Bone marrow biopsy: A piece of bone marrow is taken from the back of the hip and checked for the presence of abnormal cells.
- Lymph node biopsy: As with the bone marrow biopsy, a sample is examined for the presence of abnormal white cells. (Lymph nodes are found throughout the body and help trap and destroy viruses and bacteria.)
- Lumbar puncture (spinal tap): A sample of spinal fluid is removed and checked for the presence of abnormal cells. The presence of abnormal cells can indicate that leukemia has spread to the central nervous system.

American Blood Cancer Society

As an adult, your body will contain about 10 pints of blood. This precious fluid flows through thousands of miles of veins, arteries, and capillaries. Let's take a look at the three most-common types of cells found in the blood. First are the **red blood cells**. They transport oxygen and nutrients to the body's cells. Second are the **platelets**. They help stop bleeding and repair wounds. Last, the **white blood cells** help protect against infection by viruses and bacteria.

Figure 1. Red blood cell.

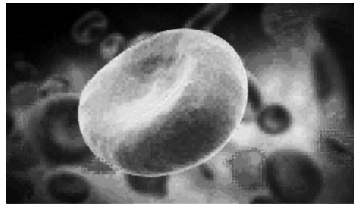


Figure 2. Platelet.

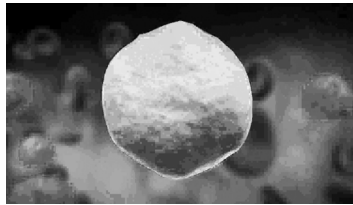
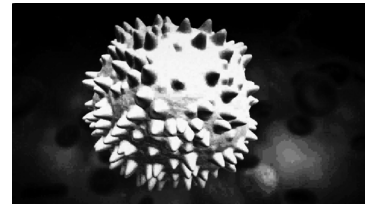
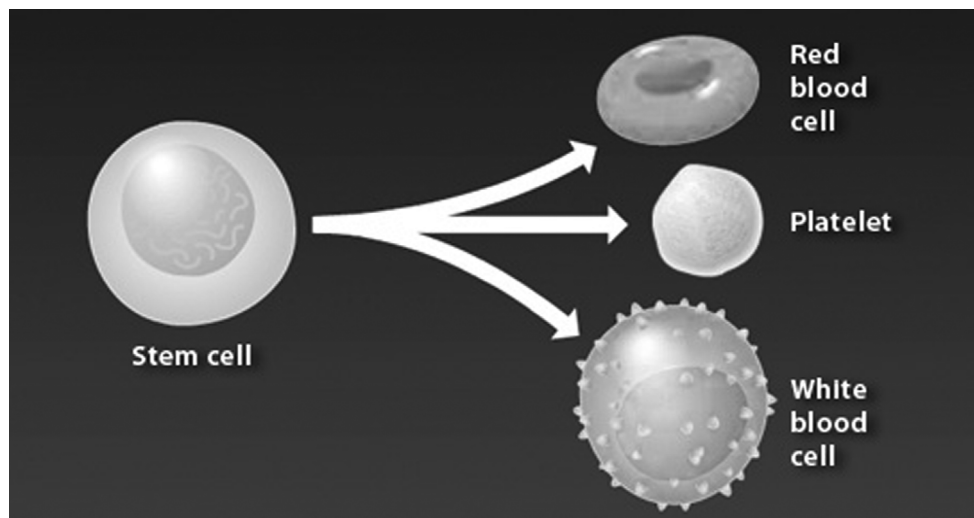


Figure 3. White blood cell.



Red blood cells, platelets, and white blood cells are not made in the blood itself. Instead, they are made in a spongy tissue called **bone marrow** found in the hollow portions of bones. The bone marrow produces cells called **stem cells**. The stem cells can divide to form red blood cells, platelets, and white blood cells, which enter the bloodstream, where they do their work.

Figure 4. A stem cell in the bone marrow can produce a red blood cell, a platelet, or a white blood cell.



American Blood Cancer Society (continued)

Leukemia and stem cells

In leukemia, a stem cell becomes mutated and begins to produce unhealthy white blood cells in large numbers. Leukemia is a cancer of the stem cells that are responsible for making white blood cells. As a result, unhealthy white blood cells are produced in great numbers and crowd out the healthy white blood cells. This also causes fewer red blood cells and platelets to be made. When healthy white blood cells encounter bacteria in the blood, they engulf and destroy them. But unhealthy white blood cells produced due to leukemia are not able to attack and destroy invading bacteria.

Figure 1. A mutated stem cell produces another mutated stem cell as well many unhealthy white blood cells.

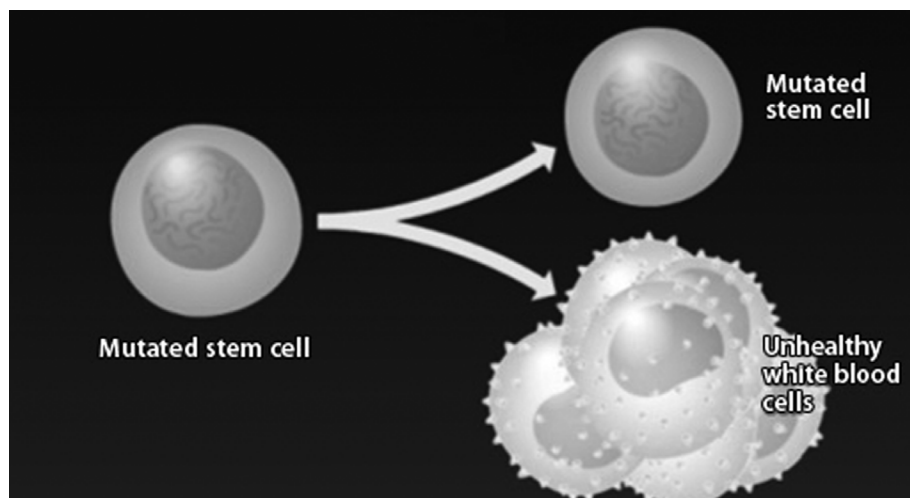


Figure 2. Healthy white blood cells engulf and destroy bacteria.

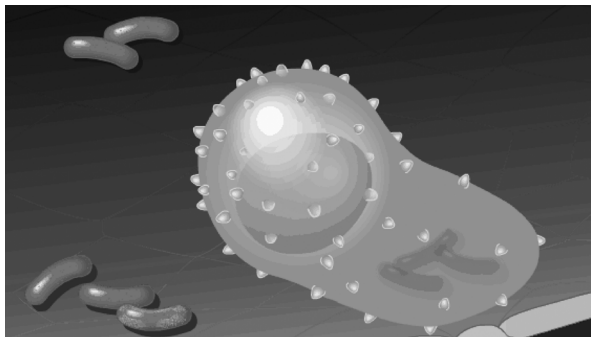


Figure 3. Unhealthy white blood cells do not engulf bacteria.



Family Karyotypes

Figure 1. Hanna. (Genetics Department, Affiliated Laboratories, Inc., Bangor, Maine)

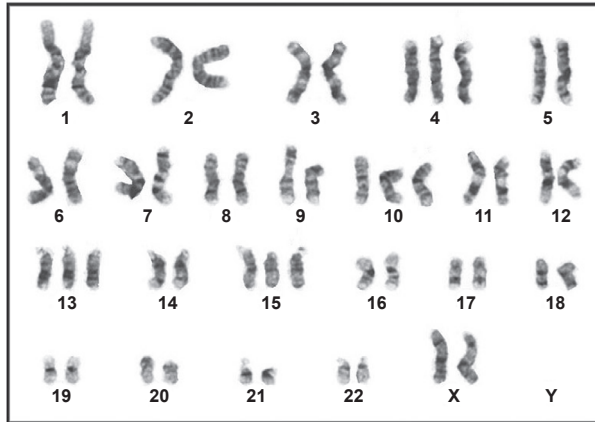


Figure 2. Kim. (Genetics Department, Affiliated Laboratories, Inc., Bangor, Maine)

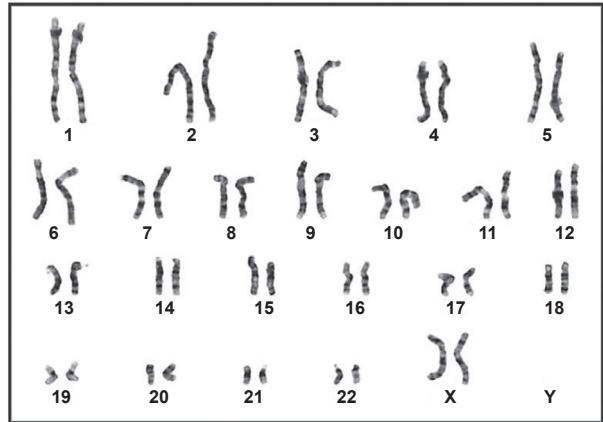


Figure 3. Rick. (Genetics Department, Affiliated Laboratories, Inc., Bangor, Maine)

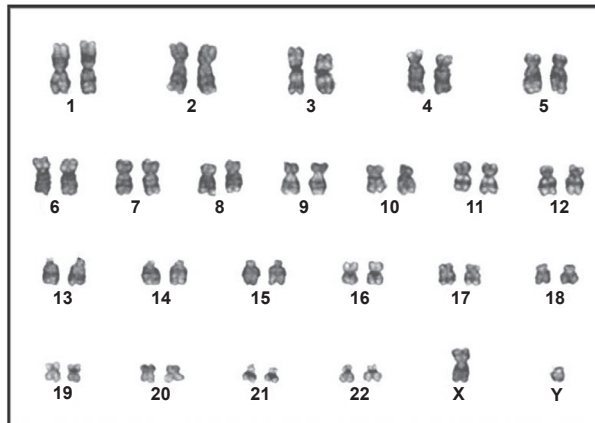
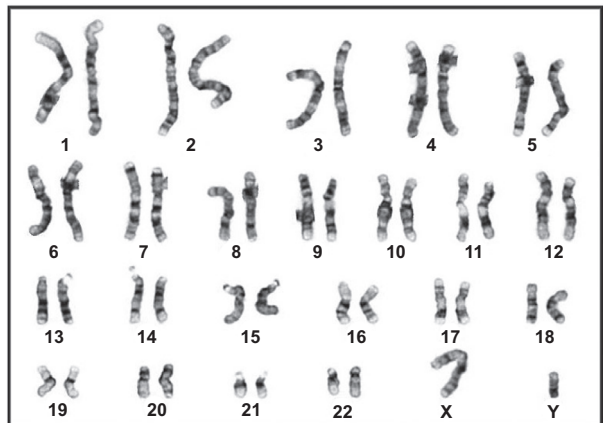


Figure 4. Jason. (Genetics Department, Affiliated Laboratories, Inc., Bangor, Maine)



Hanna's Karyotypes

Figure 1. Hanna's blood cells. (Genetics Department, Affiliated Laboratories, Inc., Bangor, Maine)

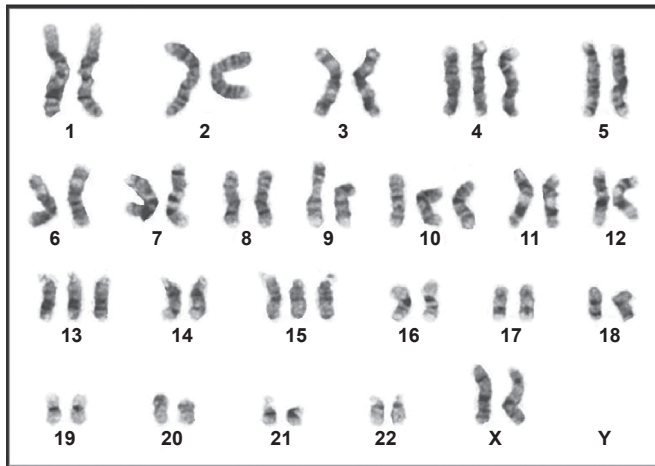


Figure 2. Hanna's hair follicle cells. (Genetics Department, Affiliated Laboratories, Inc., Bangor, Maine)

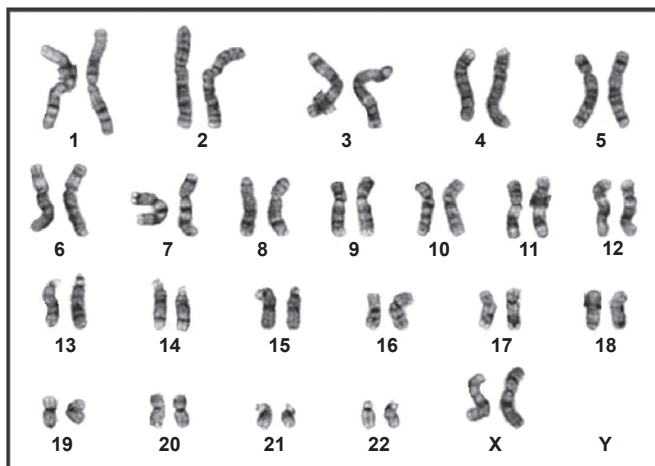
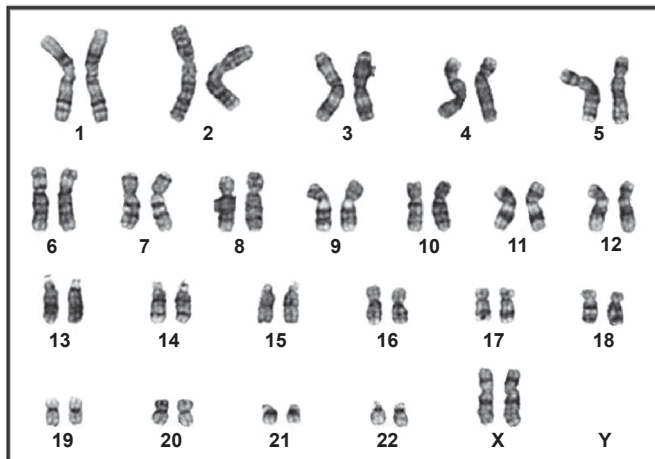


Figure 3. Hanna's cheek cells. (Genetics Department, Affiliated Laboratories, Inc., Bangor, Maine)



Changing the Dose of Drug A

Figure 1. Drug A, increased dose.

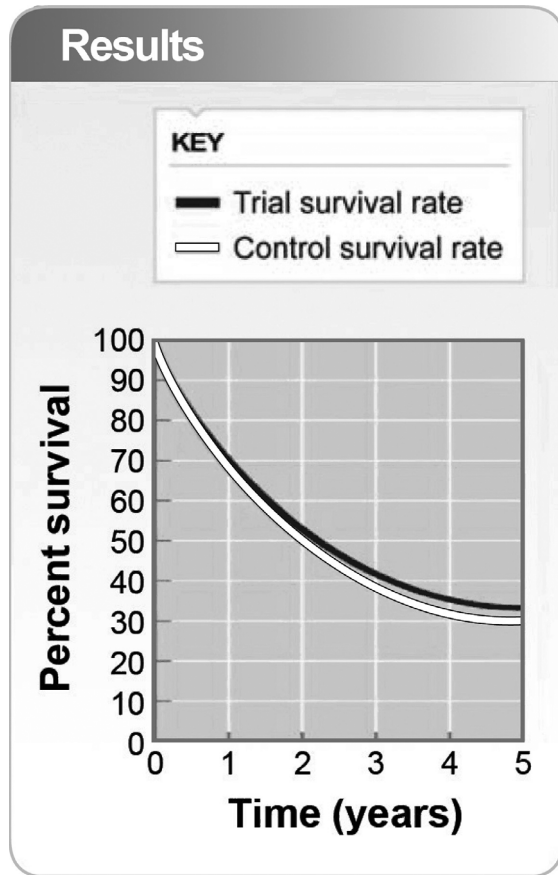
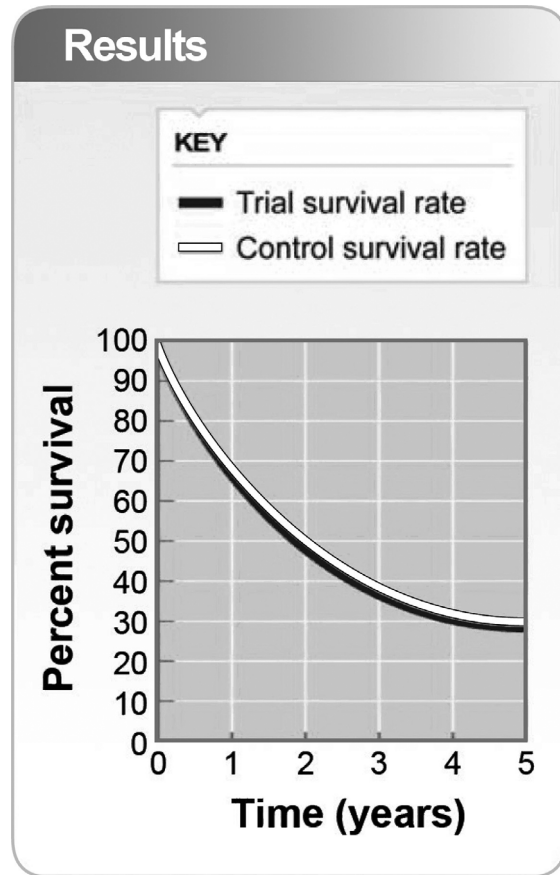


Figure 2. Drug A, decreased dose.



Changing the Dose of Drug B

Figure 1. Drug B, increased dose.

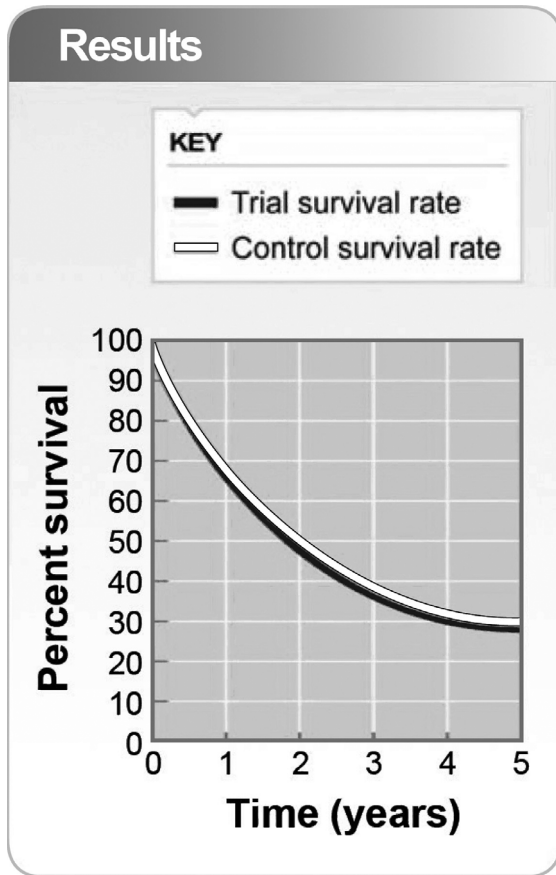
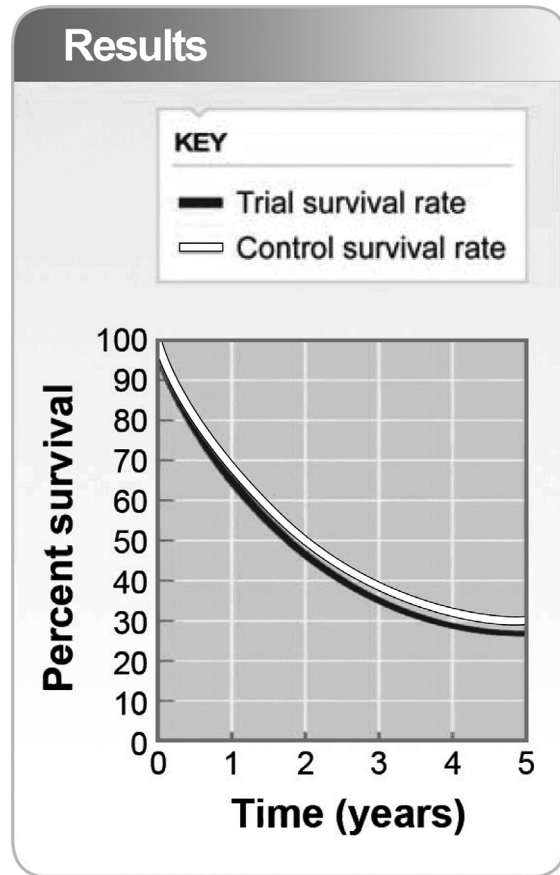


Figure 2. Drug B, decreased dose.



Changing the Dose of Drug C

Figure 1. Drug C, increased dose.

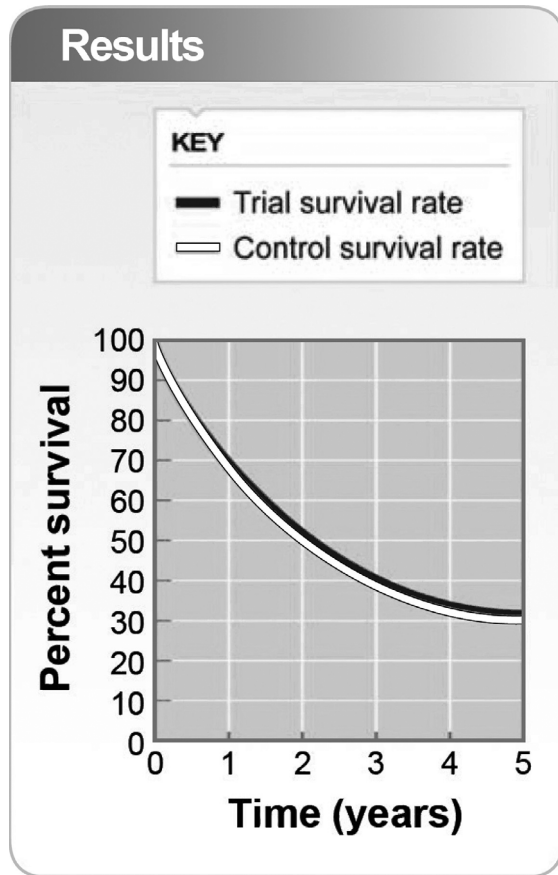
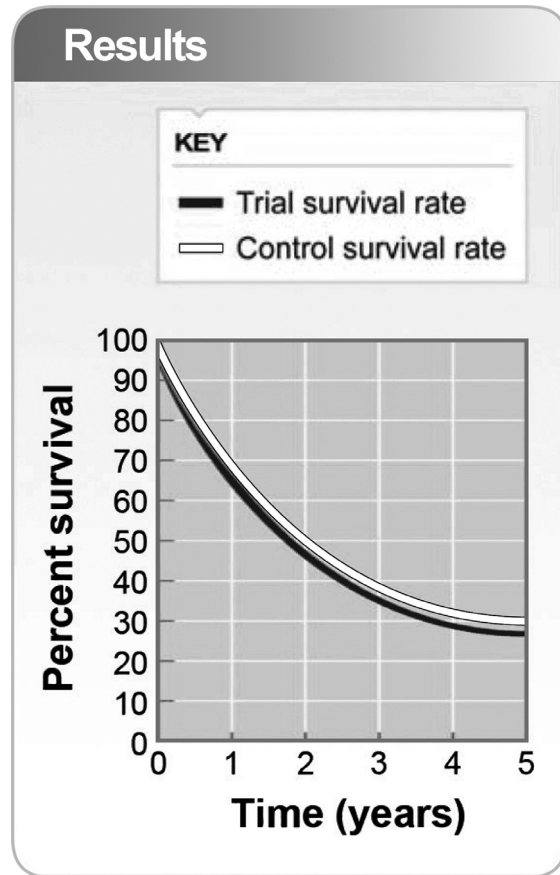
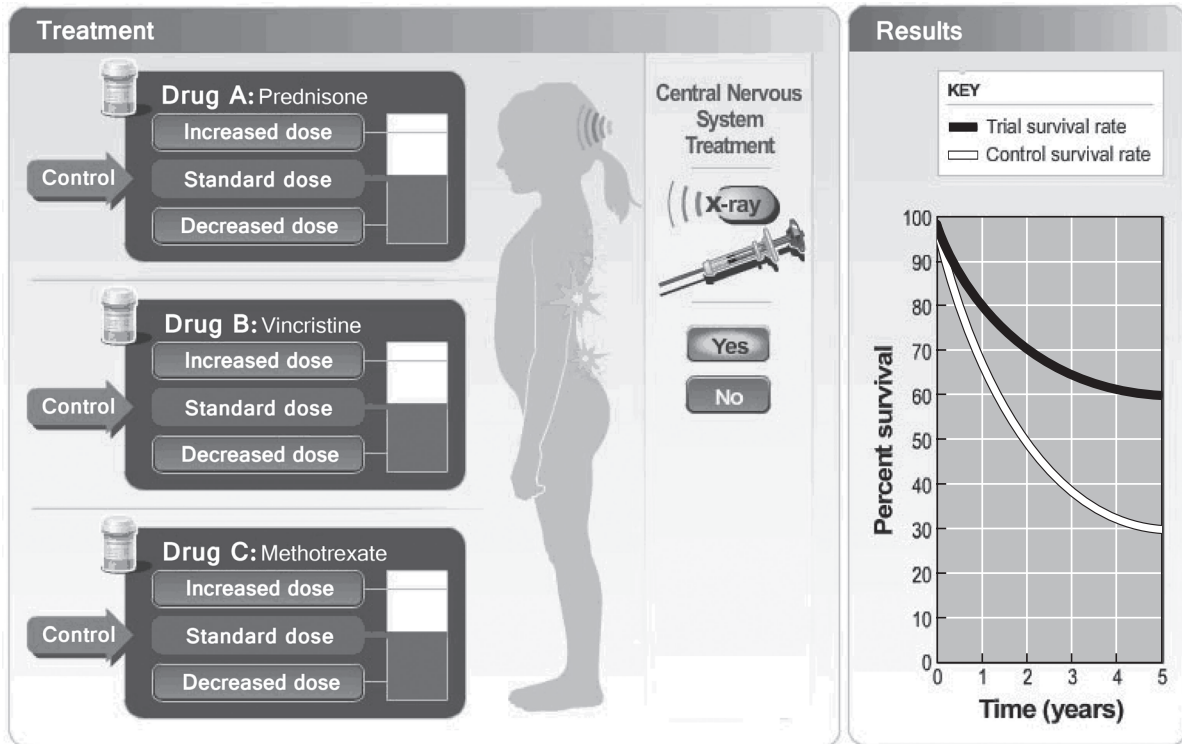


Figure 2. Drug C, decreased dose.



Central Nervous System Treatment

Figure 1. Designing a clinical trial. Left: Design of an experiment using standard doses of drugs A, B, and C with and without the central nervous system treatment. Right: Results of the experiment, where Trial is with the CNS treatment and Control is without it.



Interview with Hailey

INTERVIEWER: What was it like to go to a new high school after undergoing treatment for leukemia?

HAILEY: Being a freshman is hard already, but for me, it was even harder. After a year away, I didn't really fit in. I wasn't the same as the other kids. They were thinking about cars, cell phones, and dating. Some days, all I could think about was dragging myself to the next class without throwing up. It was strange; here I was, surrounded by 2,000 other people, and I felt more alone than I'd ever been in my entire life.

INTERVIEWER: So what happened?

HAILEY: Slowly, things started to change. I met new friends who made me laugh, and I reconnected with old friends I'd known since elementary school. My teachers helped me, too. Mr. Brinthom got me involved in theater, which made every day a new adventure. Dr. Bradley and my other teachers helped me discover a love of learning, especially science.

INTERVIEWER: Now that you're through with your treatment, what has cancer taught you?

HAILEY: Having cancer changed me. I had a new purpose. I wanted to help other teens who were going through the same thing.

INTERVIEWER: How did you go about helping teens with cancer?

HAILEY: I started delivering gift bags to a local hospital. Some people noticed and started sending me money to keep going—and I got noticed by the Leukemia and Lymphoma Society. I was even awarded the Spirit of Tom Landry Award last year by Mrs. Landry herself.

INTERVIEWER: What are your plans for the future?

HAILEY: I want to devote my life to fighting cancer as a pediatric oncologist. In the meantime, I'm helping out at events like a fashion show just for cancer patients.

INTERVIEWER: What can the rest of us do?

HAILEY: You don't have to find the cure for cancer to help someone in your own community. Sometimes, you just have to be there. Just get involved and do something—big or small.

Guidelines for the Poster

The information on your poster should be written in clear sentences in paragraph form.

Part 1: Information about the Disease

- What is the name of the disease?
- Is it a common or a rare disease?
- What are the disease symptoms?
- Who gets the disease?
- What causes the disease (and how do we know)?
- How is the disease treated?
- How might having the disease affect a person's
 - ability to participate in normal activities?
 - relationships with family, friends, classmates, and strangers?

Part 2: Summary of a Clinical Trial or Research Study

- What is the purpose of the clinical trial or research study?
- Describe how the clinical trial or research study was carried out.
- What were the results of the clinical trial or research study?
- What can you conclude from the evidence presented?

Research Study on Marfan Syndrome

Figure 1. Mice with the Marfan mutation. (© Brandon Laufenberg | iStockphoto.com)



1. Background

A drug called losartan is used to treat people with high blood pressure.

Many problems caused by Marfan syndrome, such as an enlarged aorta (blood vessel), are due to a substance that works as a signal between cells. If too many signals are sent, the aorta grows too big. When it becomes too big, it is weaker and tears more easily. Researchers wonder if a drug called losartan can block this substance and keep the aorta at the normal size. They are testing this drug first in mice with Marfan syndrome and then, they hope, in humans.

2. Study design

The study used three groups of mice:

Group 1: Healthy mice (without Marfan syndrome). They received no treatment.

Group 2: Mice with Marfan syndrome. They received no treatment.

Group 3: Mice with Marfan syndrome. They received the drug losartan.

After receiving the drug (or not) for six months, the amount of growth in the heart's aorta was measured.

Table 1. Marfan Study Results

Treatment group	Average amount of growth in aorta
Healthy mice (no drug)	0.20 millimeters
Marfan mice (no drug)	0.66 millimeters
Marfan mice (treated with drug)	0.18 millimeters

Clinical Trial on Childhood Leukemia

Figure 1. Combination drug therapy. (EyeWire)



Figure 2. Brain irradiation. (© Colleen Butler | iStockphoto.com)



1. Background

Since the 1960s, children with leukemia have had their heads X-rayed to prevent brain cancer. This treatment has worked well. Survival rates of children with leukemia have improved. Unfortunately, this treatment can also harm the ability of the brain to carry out its job.

Using more than one drug at the same time also has helped improve survival rates of children with leukemia. Researchers wondered whether the careful use of multiple drugs could keep survival rates high without using X-rays.

2. Study design

Children were assigned to one of two groups:

Group 1: Received combination drug therapy alone.

Group 2: Received combination drug therapy along with brain irradiation.

Table 1. Childhood Leukemia Study Results

Treatment group	5-year survival rate
Drug combination therapy without brain irradiation	94 percent
Drug combination therapy with brain irradiation	93 percent

Evaluation Rubric for Poster

Category	Excellent (5 points)	Good, but could be better (3 points)	Needs a lot of improvement (1 point)
Information about the disease is complete.	Each item on Part 1 of Master 5.1 is addressed.	Some items on Part 1 of Master 5.1 are not addressed.	Most items on Part 1 of Master 5.1 are not addressed.
Information about the clinical trial or research study is complete.	Each item on Part 2 of Master 5.1 is addressed.	Some items on Part 2 of Master 5.1 are not addressed.	Most items on Part 2 of Master 5.1 are not addressed.
Information on the poster is accurate.	Statements about the disease are supported by all relevant facts. Facts are accurate. Unrelated facts are not included.	Some supporting facts are missing or some supporting facts are not accurate. Some supporting facts are not relevant.	Most supporting facts are missing or they are inaccurate.
Information on the poster is clear and well written.	There are no errors in spelling or grammar.	There are a few errors in spelling or grammar.	There are many errors in spelling and grammar.
Poster design is creative and well executed.	Poster design is especially attractive, and the information is clearly presented.	Poster design is reasonably attractive, and most of the information is clearly presented.	Poster looks like it was made without much thought to an attractive and clear presentation.

Poster Score Sheet

Name: _____

Use the evaluation rubric to score the poster. Enter your score for each category on this score sheet. Include a specific reason (or reasons) for each score given.

Code number for the poster being evaluated _____

Category	Score	Reason for score (For example, if you find an inaccurate statement on the poster, list it here and include a correction.)
Information about the disease is complete.		
Information about the clinical trial or research study is complete.		
Information on the poster is accurate.		
Information on the poster is clear and well written.		
Poster design is creative and well executed.		
Total score		

What Do You Think Now?

Name: _____

1. What is a disease?
2. How do doctors tell if someone has a disease?
3. What do you think causes disease?
4. What does it mean to call a disease “rare”?

Another Letter from the Producer



Dear Principal:

I am pleased to learn that you are still interested in helping us develop our reality television show. We have been busy interviewing young people who have a rare disease, and we have selected a boy named Kevin to be the focus of the show.

Kevin has been diagnosed with a rare disease called Marfan syndrome. He is outgoing and loves sports, though recently he has had to give up playing because of his Marfan syndrome. Kevin has developed an interest in videography and has a real talent for it. He is eager to join the class and even wants to film parts of the show himself!

I will be bringing Kevin to the school sometime next month to meet you, your staff, and, most importantly, the students who will become his classmates. Of course, if you have any questions, please contact me.

Once again, thank you for your help with this project.

Sincerely,

Vincent Shifflett
Senior Producer
People Reality Productions

